GENE NEWS

A Publication of the Hawai`i Department of Health Genetics Program

Volume 5 Issue 3
July 2006

INSIDE THIS ISSUE:

GENETICS FOR YOUR PRACTICE: PUTTING THE PIECES TOGETHER

TUESDAY, OCTOBER 17th, 2006

Genetic disorders impact individuals of all ages and the impact may extend to entire families. The ability to use genetic information to improve patient care and develop prevention strategies is expanding rapidly. Healthcare providers and public health professionals should have a basic understanding of how genetic information can help improve patient care and the prevention of disease.

With the knowledge, skills, and resources gained from the conference, attendees should be able to:

- **Recognize patients with or at risk for a genetic disorder in their practice;**
- **Determine when to make appropriate referrals for genetic services;**
- **Find local genetic service providers;**
- **Identify local, regional, and national genetic resources for health care providers and families**.

The conference will also provide information about current and future innovative applications of genetics.

Who Should Attend:
This conference is targeted towards physicians, public health professionals, nurses, other allied health professionals, and students in these disciplines.

Conference brochures will be mailed in late July to those who are on our mailing list and will also be available on our website. If you need more information, please contact Pauline at pauline@hawaiigenetics.org or (808) 733-9055.

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NEIGHBOR ISLAND PEDIATRIC GENETICS CLINICS

Pediatric genetics clinics have begun at some neighbor island sites as part of our federally funded Western States Genetic Services Collaborative activities. In partnership with Hawai'i Community Genetics, the clinics are held once a month at the State District Health Offices. Clinics are currently scheduled for Hilo, Kona, Wailuku, and Lihue.

A geneticist and genetic counselor coordinate the clinics with the help of the local state social worker and/or public health nurse. The clinics that have been held so far have been successful and very rewarding. Neighbor island families seem pleased to have the option of not traveling to Oahu for their genetics consultation, especially with children who may be medically fragile.

If you would like more information about these clinics, including how to make a patient referral, please contact **Hawai'i Community Genetics at 808-973-3403**.

If you would like more information about the Western States Genetic Services Collaborative activities, please visit the website at www.westernstatesgenetics.org.

POLICY CHANGE:

Newborn Metabolic Screening for Children Older than 6 Months of Age

As of May 1, 2006, our state contracted newborn metabolic screening (NBMS) laboratory, the Oregon State Public Health Laboratory (OSPHL), announced that **it will no longer accept initial specimens for newborn metabolic screening from children older than 6 months of age.** Their laboratory standards are tailored to newborns between 0 to 6 months of age especially with our expanded NBMS panel. To ensure accurate newborn screening results, OSPHL cannot apply these standards to older infants and children. **Do not** continue to submit any initial samples to the state program for children older than six months of age.

For more information about the policy change and how to have children greater than 6 months of age screened for their initial NBMS, please call the NBMS coordinator, Christine Matsumoto at 733-9069 or go to our website at www.hawaiigenetics.org.

Coordinator's Corner

I want to welcome Sarah Scollon and Amelia Chappelle to our DOH Genetics Program. Sarah is a new graduate of the University of Michigan Genetic Counseling Program and she has experience in genetic research too. She is very enthusiastic about joining our public health genetics team and we look forward to working with her. Amelia is a genetic counseling and patient advocacy graduate student at Sarah Lawrence College. She will be interning with us this summer and we are sure to keep her busy. Please extend your aloha to them when you meet them.

Comings and Goings

It is also time to bid farewell to Holly Snyder, the genetic counselor at the Kapiolani Medical Center Fetal Diagnostic Center. She will be moving back to the mainland this summer. We wish her continued happiness and success in her future endeavors.

I enjoy hearing from our readers. Please contact me if you have further questions or comments.

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For more information, go to www.hawaiigenetics.org

Clinical M.D. Geneticist
It's All About Patients

This is the second installment of a continuing series of articles describing the various roles of genetics professionals. We hope the information will be useful for healthcare providers referring for genetic services, families who are referred for genetic services, and those considering a career in genetics.

Clinical geneticists must stay on top of the rapid advances in science and technology in genetics and apply their knowledge immediately when providing genetic services. By working with genetic counselors, nurses, nutritionists and other health care providers, they lead the team in diagnosing genetic disease. This is done by reviewing clinical symptoms, medical and family histories, laboratory tests, and current medical literature, as well as through ordering genetic tests. The goal of these genetics evaluations is to provide families with a diagnosis and the necessary information, medical management, follow-up care, and support.

There are currently 4 American Board of Medical Genetics (ABMG) certified clinical M.D. geneticists in Hawai'i. Two of the geneticists specialize in the care of pregnant women and are also board certified in obstetrics/gynecology and perinatology. The other two specialize in the care of children and adults and are also board certified in pediatrics.

Without a diagnosis, it is hard to know how to treat or potentially cure a disease, discuss potential health risks, provide medical options, or offer support resources to the individual and family. The crucial role of the clinical M.D. geneticist is to perform a genetics evaluation to diagnose and to assess the risk for a genetic disorder.

After medical school, most M.D. geneticists do a residency in a specialty like pediatrics, obstetrics, or internal medicine before beginning their two year fellowship/residency in medical genetics. Some medical doctors can enter a 48 month medical genetics residency program or a combined specialty and genetics residency program. Clinical M.D. geneticists are certified by the ABMG by taking the clinical geneticist examination. Additional subspecialty genetics certifications (biochemical, molecular or cytogenetics) can also be obtained from the ABMG if the candidate meets the requirements and passes the examination for the subspecialty.

If you are interested in more information about this profession, please go to:

American Society of Human Genetics:
www.ashg.org
American College of Medical Genetics:
www.acmg.net

Genetic diseases affect people of all ages from fetuses to children to adults. Listed below are some of the common reasons to refer a person for a genetics evaluation.

- Medical problems of the developing baby detected by prenatal screening or testing.
- Harmful exposures during pregnancy including alcohol and prescription or recreational drugs.
- Birth defects such as heart problems, structural brain abnormalities, and cleft lip and palate.
- Mental retardation or developmental delays where the person does not reach developmental milestones on time or they do not function developmentally, intellectually, socially, or behaviorally as expected for age.
- Changes in body chemistry such as extremely high and low protein, fat, or sugar levels in the blood.
- Sensory impairments like vision or hearing problems.
- Family history of a hereditary disease or cancer.

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Healthy Mothers, Healthy Babies Conference

The Hawai'i Genetics Program staff presented at the "New Frontiers in Maternal and Infant Health Care: Ethics, Outcomes, and Practices in the 21st Century Conference" held on June 7th and 8th. The conference was sponsored by the Healthy Mothers, Healthy Babies Coalition of Hawai'i and the Department of Obstetrics, Gynecology and Women's Health, John A. Burns School of Medicine. More than 175 health professionals, students, and policy leaders attended.

Notable speakers included Dr. Hani Atrash, the Associate Director for Program Development at the CDC National Center on Birth Defects and Developmental Disabilities, and Professor George Annas, a faculty member at the Department of Health Law, Bioethics, and Human Rights at Boston University.

The Genetics Program staff presented a session that covered current pre- and post-natal genetic testing techniques with a focus on the ethical, legal, and social issues, as well as original research conducted by the Genetics Program. The highlight of the session was the use of an audience response system (similar to the system used on *America's Funniest Home Videos* for voting) to tally participant responses to different ethical case studies.

Judging from the audience responses during and after the session, the attendees thoroughly enjoyed the presentation and were thrilled by the use of the audience response system technology. Thank you to the Healthy Mothers, Healthy Babies Coalition for allowing us to participate in the conference!